The Children's
Medical Research
Foundation, Inc.®

KirbyGram®

December 2020

Our goal is to create awareness of Sanfilippo Syndrome and other neuro-genetic disorders, fund medical research and find a cure.

Dear Friends,

What a year…

For Brad and me, a year of firsts without our beloved Kirby. Never having more freedom, yet never more confined. And throughout, there were those birthdays and holidays that challenged us to see past our sadness and find the "happy." Kirby's legacy was perfectly clear and enabled us to do just that.

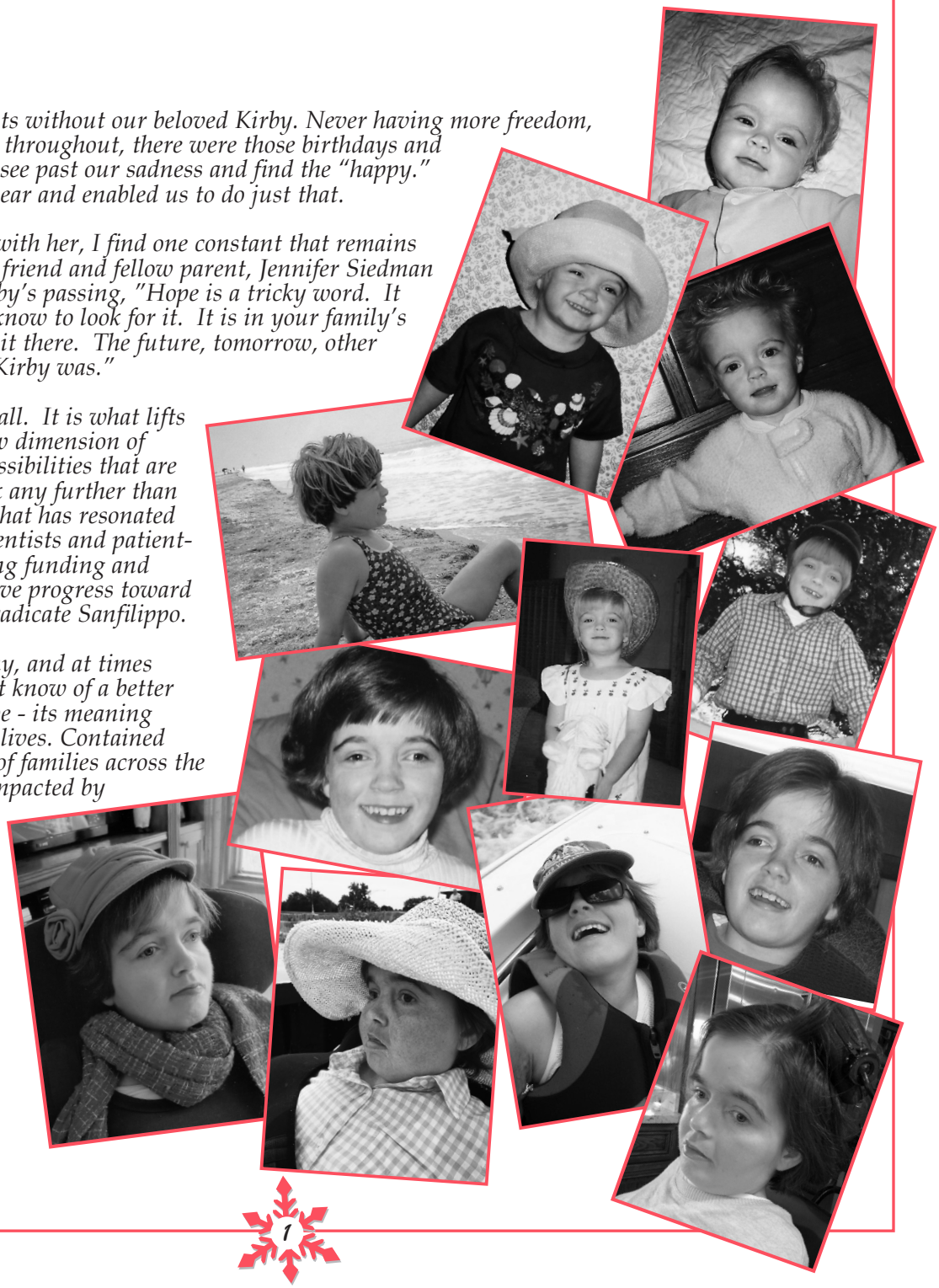
Now, in reflecting on our life with her, I find one constant that remains and has endured – hope. As a friend and fellow parent, Jennifer Siedman described after learning of Kirby's passing, "Hope is a tricky word. It is fluid. Enduring when you know to look for it. It is in your family's hearts, even today. Kirby put it there. The future, tomorrow, other families' hope is here because Kirby was."

I believe that hope transcends all. It is what lifts you and harnesses a whole new dimension of thought, filled with endless possibilities that are empowering. We need not look any further than the relentless hope of parents that has resonated with an alliance of donors, scientists and patient-focused biotech leaders, creating funding and inspiring resolve and innovative progress toward the community's mission to eradicate Sanfilippo.

Our challenges have been many, and at times for some, seem endless. I don't know of a better time than now to dwell on hope - its meaning and commonality in all of our lives. Contained within these pages are stories of families across the world whose lives have been impacted by Sanfilippo. Their children's faces define joy and hope in its most innocent and purest form. Whether they are able to hold their children in their arms or only in their hearts, their stories of hope inspire. I hope you feel the same.

Brad and I wish you a happy and hope-filled holiday season.

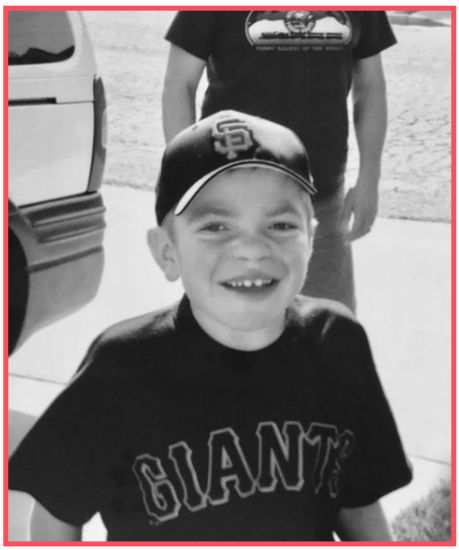
—Sue and Brad Wilson



Stories of Hope

Andrew & Nathanael

Although the years have been many, I have never forgotten the comfort Kirby's face brought to Rob and me the day we found the Foundation on the Internet. When Andrew was being diagnosed and no one we knew had heard of MPS, your website and your angel brought us such comfort. Our Andrew was not alone.



In the years since that moment, we had to say goodbye to Andrew when he passed away at age ten in 2005. We also welcomed our son, Nathanael, and walked with him on his journey with MPS until he passed away in 2016 at age nine. Ironically, I do not think I ever knew the true meaning of hope or joy until those two angels came into my life. Before finding out Andrew had MPS, I was a busy person who thought a career and a perfect family was the thing for me. I thank God everyday for all four of my sons - but especially Andrew and Nathanael - who took our family on a completely different journey. A journey that is often hard to put words to because it changed all my values and taught me that the most important thing in life was not accomplishments or contributions, but moments. Every single moment - the moments I spent cuddling, diapering, reading to, laughing with, comforting, playing with, spoon-feeding, wheelchair-transferring, bathing, and taking my boys out to enjoy their community - those were the most meaningful moments of my entire life.

I spread more joy and hope now than I did before I met them because through them and our family's MPS journey, I finally learned the meaning of life and the value of every moment we are given on earth. I could have four healthy, living sons today, and there was a time when I would have made a deal with the devil to make that dream a reality. Yet because of Andrew and Nathanael, I now completely understand why it happened the way it did. I still hear from people of all ages who spent time with our angels, and every one of their lives is different because of it.

We need to cure MPS not only because everyone deserves a long and pain-free life, but because these angels need to be on earth longer to teach the rest of us.

Wendy Coffey-Slattery
West Little Rock, Arkansas

Matthew

Matthew is 20 years old with Sanfilippo B and is in his last year of high school. He has brought a tremendous amount of love to all his family, friends, teachers, aides, and random people that are lucky enough to get a big Matthew bear hug upon meeting him.

I have watched him gain skills as a young boy and experienced his playful, loving, mischievous and joyful personality. I have watched him lose all his verbal and cognitive skills, but his loving joyful nature has always remained. No matter what the day brings for Matthew, whether it be seizures, sickness or other complications of Sanfilippo Syndrome, he is still smiling through it all. He loves balls, books, balloons and swimming and is happy to just hang out with us.

We hope for many more good years for Matthew and feel blessed that he came into our lives.

Kathy Buckley
Saratoga Springs, New York

Sydney

How does one continue to hope when what you hoped and prayed for for so long is no longer a possibility? It is a daily struggle, but hope continues to give us the strength to see the possibilities and experiences that await us in life. Sydney's memory is what gives us the strength to continue to hope. She was all about life.

Sydney was an absolutely joyous spirit. Her smile and laughter were infectious. She literally laughed, sang and danced her way through life. Our home was filled with the sounds of laughter, chatter and music. She also was so strong and unbelievably compassionate for someone so young. She never complained about her declining health. She had a sixth sense about other people. She would be the first one to notice if someone else was sad or in pain. She would go and try to comfort them with hugs or words. It was always so unbelievable how she just knew. So many times we would find her trying to "mother" someone who was sad or hurt.

Her life was an incredible journey filled with joy, laughter, pain, sorrow and hope. We felt like we were in combat for 20 years and our foe was Sanfilippo Syndrome and Sydney was its hostage. Despite this, we would never take back a moment. Sydney was here for a purpose and that was to show us what unconditional love is and what true inner beauty and strength look like. The world is better place because of the laughter and love she shared with all.

We continue to hope for a cure. We continue to hope that other families have the strength and support they need to fight the battle. We continue to hope that everyone enjoys every day and the possibilities that each day brings. Most importantly, we hope that Sydney's courage and strength encouraged others to be brave and to live life to its fullest.

"Do not think we know our own strength until we see how strong our love makes us."
—Leigh Standley

Theresa Moff
Williamsport, Pennsylvania

Eliza

Eliza, that's her. That's my girl. Lover of Dora the Explorer, Peppa Pig and a day at the beach. Her smile lights up my heart. There is not much her dad, brother and I won't do to try and conjure that magic. I'm sure we look like goofballs from the outside, but that's ok. Our children with Sanfilippo have a special way of grounding us to the here and now.

While this is a disease of perpetual trauma with the loss of child after child, we take hope in what lies ahead. We are comforted by a community drawn to action that continues working to change the future. We are ever thankful to the Wilsons and Kirby for their decades of work and inspiration.

Cara O'Neill
Columbia, South Carolina

Iñaki

Iñaki's eyes radiate life, goodness, tenderness. He knows no bad feelings and finds much satisfaction in comforting people, especially if they are children, and he always makes sure that everyone around him knows how much he loves them.

My son is, above all, a child full of light. I was not able to conceive, but God blessed me with just one chance of getting pregnant. Iñaki was born in 2010 after fertility and implantation treatment. Two years later, in 2012, he was diagnosed with the rare and fatal Sanfilippo Syndrome.

Is it possible to maintain HOPE when you know that you have had "the bad luck" of giving life to a child born with a cruel, painful and deadly disease? When your lives are going to always be under the threat of death? This is a question that has been going back and forth in my head since 2012. I have gone through all possible emotional stages in these eight years, and no, it has not always been possible to maintain HOPE, at least not without a strong desire and willingness to maintain it, to work for it. HOPE and FAITH are not things that just fall from the sky. Hope is built every single day; it is a decision and a constant exercise. The same thing happens with faith, and to me, they go hand in hand.

On this path, I have had to learn a lot, a lot more than in 10 lifetimes. My strength is constantly being tested, and I confess that many more times than I like to admit, I have thought that I just couldn´t make it any longer; I have also felt a loss of faith. But it is precisely in those dark moments that a decision that I made years ago becomes stronger: to spend the rest of my life working not only for the health and life of my deeply beloved only son, but also for his happiness, so I could always see that smile on his face that illuminates everything around him.

Iñaki has no one else but me; the man who was supposed to be his father, my ex-husband, left us with the diagnosis. It's not about me, it's about my son. It's about him living every day of his life feeling confident and absolutely loved. It is not about living, as I have sometimes been told, without accepting a painful reality. No, it is not that; it is learning how to live with that painful reality, but not allowing that pain to be the axis of your life and that of your child. And here comes what I could say has been the most important and firm decision that I have made for both of us and which I exercise EVERY SINGLE DAY: TO LIVE WITH FAITH and, therefore, LIVE WITH HOPE. I will never stop fighting for his life, but I know that whatever happens, Iñaki's life will have been beautiful and meaningful.

Janette Ojeda
Barcelona, Spain

Rhianna

I am honored to share our story along with those of other families in the KirbyGram, grateful that Kirby's legacy continues to generate awareness and hope. My dearest Rhianna walks with me every day, in spirit and in treasured memories. I think back to 1995 when Sanfilippo arrived on our doorstep with only one researcher worldwide and the prediction of a treatment/cure not likely in my lifetime. The future for Rhianna and us as a family was devastated. The only way to stay strong and cope was with HOPE. Hope in fundraising, in finding a cure, in beating the odds, hope that Rhianna would walk a different path than the one expected. In a few short months, friends wanted to help find a cure, and our fundraising became known as Rhianna's HOPE. The repetition of the word Hope kept us strong and focused.

Rhianna did exceed all expectations: she painted, turned pages, and attended to books, rode a bus to school, pedaled her own bike and both rode horses and swam with dolphins for many years. She had wonderful friendships and maintained her cognitive awareness to the very end of her unexpected 23 years. Throughout her journey, despite all that Sanfilippo had taken from her, Rhianna's strength never wavered. This was clear in her amazing smile, infectious laugh and joyous spirit.

I remain truly grateful to all who supported the MPS grass root efforts, pounding the pavement, fundraising and directing money to the best resources for a cure. It has been decades of hard work by MPS families, researchers and doctors. I miss Rhianna in every moment. I continue to see the same wonderful characteristics in the children out there who are battling Sanfilippo and the many types of MPS. Now with the advances in medical science and technology, along with worldwide Internet communication, there can be a firm belief that families will have a cure. Our work is not done, so I embrace hope again…hope that our stories encourage donations, hope that there is a new path for every child with MPS, one that gives them many years to share their beautiful spirits.

Love and hugs to all.

Cynthia Logan
Plainville, Connecticut

Lucas

Our Lucas touched everyone he came in contact with. All remember his smile – that contagious smile and his ability to smile despite limitations and pain. He started and ended every day with a smile. It will always be our reminder that life is what we make of it.

My hope is that despite the inevitable ending, we hold strong to the lessons he taught us. Live and love strong. Follow his lead and stay courageous and positive, and remain in the moments of here and now. There are no second chances.

Stacey Montgomery
Cedar Rapids, Iowa

Kimberly

Our Kimberly was truly an angel sent to fill our hearts with love and to teach us many lifelong lessons. Kim was such a happy, jubilant little girl. She had the spirit of Pippy Longstocking, the sweetness of Grover from Sesame Street, and the happy-go-lucky manner of Tigger. Upon meeting Tigger at Disney World (one of her favorite Disney characters), she became shy. When Tigger walked away from us, she smiled broadly and laughed and laughed.

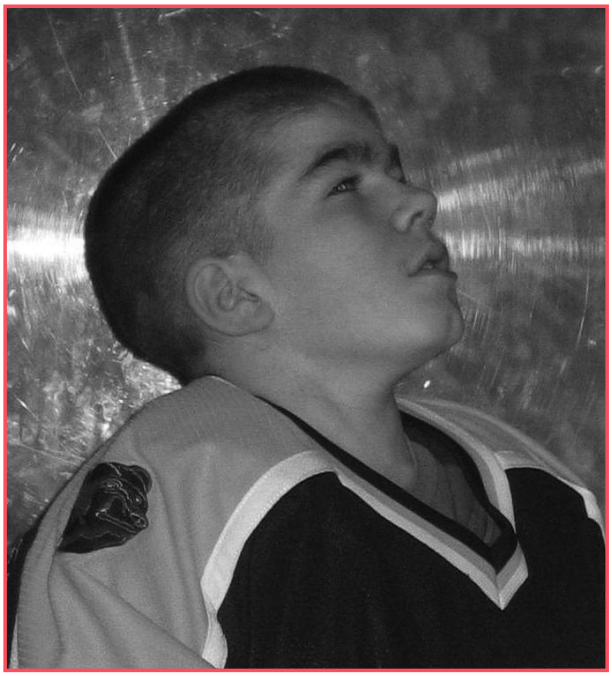
When Kim was diagnosed with Sanfilippo (not until the age of eight), we were unsure of what was ahead of us. We experienced the many facets of the disorder during her life and were told to hope for treatments and programs to support her. We tried to be prepared for each phase; all the while, she was teaching us patience and gratitude and enriching our understanding of special needs children and the special needs community. Because of Kimberly, two of her babysitters have become special education teachers and told us that they attributed their desires for this profession to her. She enriched and blessed our lives for 26 years.

Dear Sanfilippo families, we see you and honor you. It was not easy, but we learned to live in the moment. Tomorrow, next week and next year didn't matter right then; we had to appreciate all the time we had. Let your hopes not your hurts direct your future. It's been said that hope is love on a mission, and even in this turmoil of 2020, remember, "Hope is not cancelled." The work that The Children's Medical Research Foundation and other families' foundations are doing can give us all hope. We salute you and send you our hope for your children.

Betsy Fowler
Shelby Township, Michigan

Ben

Hope is my North Star.



It seems that loving a child with Sanfilippo has much in common with a trip to Mars. I wish I had thought of this title, but alas it is a quote from the Netflix series Away, a fictional story about a trip to Mars.

The North Star is the sky's anchor. Its bright glow helps travelers determine their direction and reach their goal. Hope has been my North Star since the day Ben was diagnosed, and it continues to guide me even now that he is gone. With a vivacious, cow-loving 15 month old on my lap, it was a hope for treatment that initially anchored me and helped conquer the overwhelming anticipatory grief that I was feeling for all the things that would never be. And hope was most definitely the fuel that drove my resolve as his mother to provide him with every opportunity to reach his greatest potential, even as he was losing his abilities and Sanfilippo was beating him. The pure, simple hope of hearing a sound, watching his lips curl in a smile or having his eyes lock with mine in that way that said "I love you, Mom" made even our most challenging days better. Hope most certainly anchored me on the day Ben died. On that day as we turned off the ventilator, my hopes were for his peace, for my other children's hearts to heal, for mine to never forget, for the courage and determination I knew I would need in the coming weeks. I know it sounds odd (except to those who know), but even the impossible is better when hope is part of the equation. You see, like the North Star, hope is visible no matter where you are. You just need to remember to look for it.

PS. The same series produced another good quote – one that most certainly applies to the effort of so many families, like mine and like the Wilsons, to drive science and bring treatments - "The hope that we might succeed is enough to keep us trying."

Jennifer Siedman
Wellesley, Massachusetts

Jonah

Season's Greetings. I'm Jill Wood, Mother to Jonah Weishaar, a cheeky little chap who loves to play jokes on his parents. Jonah could tell you knock-knock jokes all day long, and he loves hiding and surprising his friends and family. His laugh is so infectious that you can't be angry when he scares the pants off of you. Jonah had the misfortune of receiving two bad copies of a gene from his parents that caused him to have Sanfilippo syndrome type C. Fortunately, Jonah had an astute pediatrician who recognized the signs of Sanfilippo when he was only one year old. This gave us the chance to fight Jonah's fate. At that point, Jonah was making his milestones; to us he was perfect in every way. We didn't know that his ears were so full of fluid that he could barely hear. We were able to address his hearing during his formative years. Jonah qualified for special early education services and started receiving speech therapy when he was two years old. Jonah is now 12; he can still speak full sentences and is still learning new things. He has defied his fate in every way.

My husband and I are extremely lucky, and we haven't taken our blessings for granted. We formed a not-for-profit to raise funds for gene therapy to treat Jonah's version of Sanfilippo. We are now planning the first-ever gene therapy clinical trial for Sanfilippo type C. We're about two years out from giving Jonah a treatment. My hope is that Jonah can maintain his skills for a few years longer. If he can, Jonah could be the first child with Sanfilippo type C to live a long life. That's my hope.

Jill Wood
Brooklyn, New York

Elisa

Everyone has a story to tell. Ours is one of faith, family, sacrifice and hope. Ours started out with three beautiful children, all of whom we thought were healthy until one day, in 1998, we were given the unthinkable news that our youngest daughter, Elisa, then aged four, had Sanfilippo Syndrome. This diagnosis, at first, left us hollow, helpless, devastated and so much more. We were told to take our daughter home and enjoy her every day, as there was no treatment and very little research worldwide to find a cure. We were given no hope. We then met the Wilsons - Brad, Sue, Maggie and Kirby. We were inspired. We joined them in their efforts to expand fundraising to support research in Canada and work together to help find a cure. The Sanfilippo Children's Research Foundation, "A Life For Elisa," was created, and hope for us was born.

We are a family of faith. We believe in a God who created Elisa for a purpose and gifted her to us. We knew he would walk beside us, through the peaks and valleys on our long journey ahead of us. Hope is the confident expectation of what we wish for and desire. One cannot have hope without faith that the future will be better, where we believe in something that we cannot see or have not yet experienced. For the twenty-two years of Elisa's life, we lived each day with hope that a cure, even a treatment, would be found, and hopefully in Elisa's lifetime.

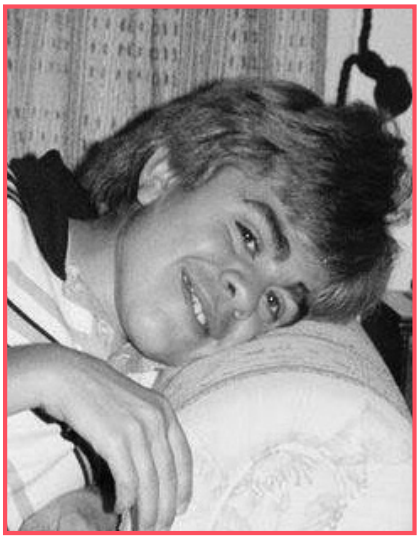
Hope is contagious. For over two decades, we have been surrounded by people who have encouraged, inspired and supported our efforts, sharing the same hope of a cure, helping us make huge strides in moving research forward, thus bringing us one step closer to that reality.

Not all stories have happy endings, but we believe ours will. Although our sweet Elisa passed away October 31, 2016, at the age of 22, our story of hope continues, as encouraging research exists in labs worldwide as a result of donations given from generous donors over the years. Our story is not over yet. The last chapter where we can say a cure has been found and a parent given the diagnosis of Sanfilippo Syndrome for their child has the reality of a treatment is yet to be written. Until then, may the flame of hope our kids have lit never be extinguished, and may it continue to make all the difference for children diagnosed today and tomorrow. Hope lives on.

Elisabeth Linton
Toronto, Canada

Jesse

2020 has been a challenging year for all of us, I think. Our Jesse, who is 25 years young with Sanfilippo Type A, has been resilient and overcomes everything that Sanfilippo seems to throw at him. This fall is starting off more challenging with what seems to be depression for Jesse, with all his resources still closed. We hope his adult program will open again soon.



Jesse has taught us so much more about unconditional love, more than our average lives could ever have taught us, even with Jesus at the center. Every tough challenge has been met with joy from Jesse, especially when we have found ways to overcome the issue. Fortunately, we have resolved most issues, and he has returned to his happy, smiling self again each time with no thought of the past. His love for everything around him is endless and so tolerant.

We have been truly blessed by his presence in our lives, even though seasons in this life have been hard and challenging at times. But even before Sanfilippo, we had challenging seasons in life in other ways. With Sanfilippo, when we overcome those challenges, the rewards are obvious, and we see the wins with the real life of this special young man God has given us for however long. It is our privilege and honor to care for and enjoy his life to the best of our ability until he returns to his forever home. We will pursue every day to be the best he and we can be.

Over the years we have followed and advocated for research to help clear substrate that builds up in Jesse and others like him. We have contributed to research that has funded many studies and trials, and yet we still have no cure. BUT, many are helping with quality of life. For that reason, we have hope that the studies we do now will give children afflicted with Sanfilippo a chance at a better quality of life until a cure is found. Never give up hope, but live your best each day for today!

Love to all our families and praying for a better 2021 for us all.

In Christ's love,

The Taormino Family, Patty, Jack and Jesse
Baltimore, Maryland

Isla and Jude

In the seven years since my beautiful children, Isla and Jude, were diagnosed with Sanfilippo, the one thing that has sustained me each and every day is hope. In the early days, I was warned by the doctors that I shouldn't have "false hope," but I have come to know there is no such thing - all hope is real. I think they thought my hope was simple, that my kids would be cured, make complete recoveries and live long and healthy lives. But hope comes in many forms. Hope is not one-dimensional. It is complex and multi-faceted. It gets us through the long days and the short years. It is enduring and it is unwavering.

Over the years my hopes have changed. Once I hoped for Isla and Jude to develop enough cognition and language that I would know who they were before I lost them. Now that Isla's words are all but gone, my hope is we might communicate through other means - with our eyes, through our touch. Once I hoped we would be able to prolong their lives, now I hope to alleviate their suffering in the short years they have. And when the time comes to say goodbye, my hope is we will be ready.

I wrote this piece about hope and what it means to me, and we turned it into a video clip featuring Sanfilippo parents. It resonated with others who are living this life, because sometimes hope is all we have.

Ode to Hope

Sometimes hope is all we have

Hope that children with Sanfilippo might face a different future to the one handed to them on diagnosis

Hope that something might be found in their lifetime to help alleviate their suffering

Hope that future generations are spared the heartache that we have today

Hope that my children are seen and loved and valued

Hope that I will see my children laugh and smile for many years to come

Hope that my children's lives are not lived in vain

Hope that the strides being made in research can alter the course of this disease

Hope that treatments we're exploring can bring us closer to a cure

Hope that every day is progress and tomorrow will be better

Hope that you will stand beside us through the good times and the bad

Hope that one day we will have a world without Sanfilippo

~ Megan Donnell
Sydney, Australia

Hope in Research

The Foundation is thrilled to announce its two-year award to fund a postdoctoral fellowship: Gene Therapy for Sanfilippo Syndrome at the Research Institute for Children's Health at Case Western Reserve University in Cleveland, Ohio, under the leadership of Dr. Mitchell Drumm.

Dr. Drumm earned his doctoral degree in the laboratory of Francis Collins, M.D., PhD, with whom he co-discovered the gene that causes cystic fibrosis (CF). Dr. Drumm has been involved in the therapeutic development processes for cystic fibrosis ever since and in 2015 launched the Research Institute for Children's Health with the philosophy that one take the successful strategies learned in CF and apply them to other genetic disorders, moving more quickly and efficiently by not reinventing the wheel. Dr. Drumm is excited to engage in gene therapy for Sanfilippo because of the transformative changes in genetic therapy technology now available. His lab and colleagues have developed a pipeline approach to move therapeutics to the clinic in other rare, genetic disorders using gene-based therapies.

Dr. Drumm's implementation of laboratory-to-clinic research programs for rare, genetic disorders patterned after the CF approach is exciting and now provides a fresh look at gene therapy for Sanfilippo, revitalizing hope for families and children afflicted.

Dr. Drumm has described for us that his laboratory has created mouse models that allow easier and stronger detection of successful steps in the gene therapy process. He says that current technologies don't allow us to look for the enzyme in individual cells of the body, or how long it lasts. The cells of the mice they've developed actually light up if the gene therapy approach reaches them, making them straightforward to find. Dr. Drumm explains "Our mice, that let us see where the gene therapy particles go, how many cells they enter, and how long they last, will provide a guide when we compare different gene therapy approaches in the Sanfilippo mouse that the Foundation had the foresight to fund. We have teamed up with AAV gene therapy experts as the first approach we will try, but are set up to evaluate any system one wishes to try, or even combinations of systems. The support from the Foundation will allow a talented postdoctoral fellow in my lab, Karen Schelde, PhD, who has extensive experience with mice in this regard, to test the gene therapy strategies." Dr. Drumm is very excited for his lab to work on Sanfilippo. "Quite simply, this is why the Research Institute for Children's Health exists," he says. "Our researchers may be scientists, but they are also parents."

Dr. Tim Miller, Co-Founder, President and CEO of Forge Biologics in Cleveland, Ohio, introduced Dr. Drumm to Sue and Brad Wilson. The Wilsons and Dr. Miller first met when he joined forces with the Sanfilippo community in 2012 and founded Abeona Therapeutics.

On a personal note, Brad and I are excited by the Foundation's opportunity to include Sanfilippo in Dr. Drumm's work said Sue Wilson. It circles back to our beginning - funding the development of a mouse model and including Sanfilippo in the most promising research, which included AAV gene therapy research – utilizing both with new "tools." And now, because of Tim's introduction, we have the honor of working with a gentleman, like Tim, who wears his compassion for children afflicted on his sleeve. Their passion and morals and the integrity of their work exemplify all the Foundation's values of being, sharing and enduring hope. Quite simply, it just feels right in our hearts. Kirby is smiling.

Faces of Hope





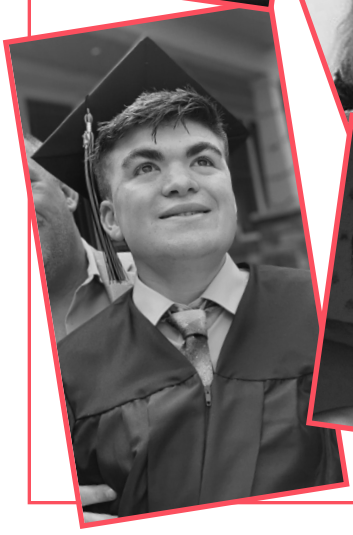


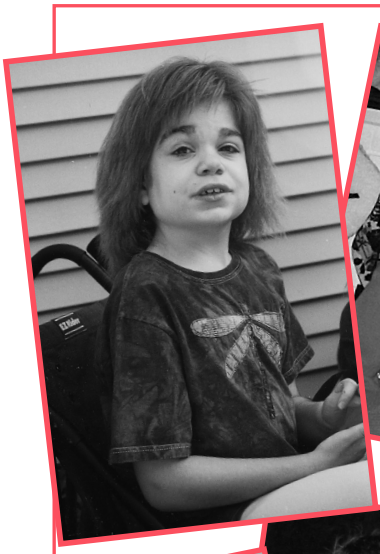


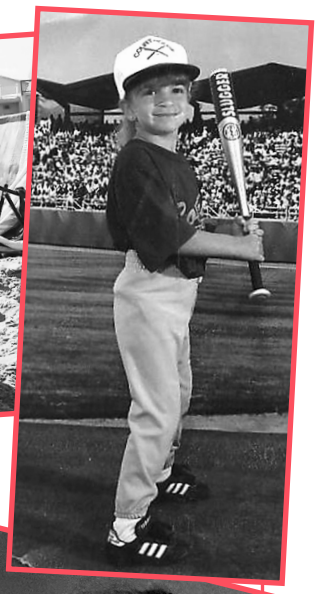


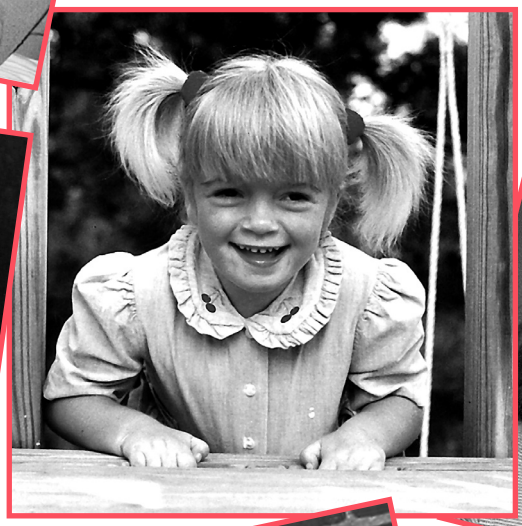


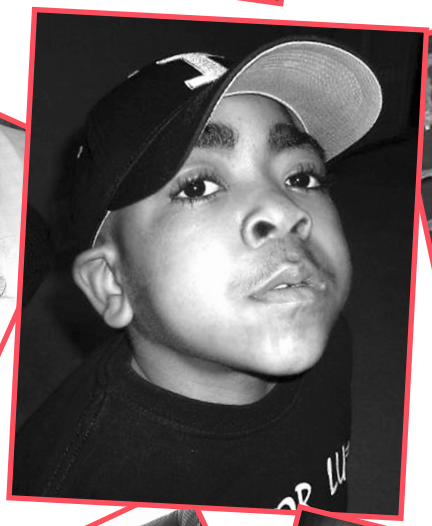




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